ABSTRACT OF THE DISCLOSURE

Samples are tested for mutations in the BRCA1 gene using a hierarchical approach. First, each sample is amplified in one or more multiplex PCR amplification reactions. Each multiplex PCR reaction produces a mixture of amplified fragments. The sizes and amounts of these fragments are evaluated and compared to standard values reflecting the sizes and amounts of fragments produced when the same multiplex amplification is performed on the wild-type BRCA1 gene. Differences between the observed fragment sizes and/or amounts and those for the wild-type gene are indicative of a mutation with the BRCA1 gene of the sample. Next, one or more of the exons of the BRCA1 gene are sequenced, preferably only for those samples where no mutation was detected by analysis of the multiplex PCR fragments. The sequencing procedure can be performed by amplification and sequencing of the multiplex amplification mixture.